

The process of how mothers with down's syndrome children learn about the diagnosis: A qualitative research

Informing about down syndrome diagnose to mothers

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Abstract

Aim: Healthcare providers have an important role in notifying mothers about their children's handicap, in other words, the bad news. The mothers' acceptance of the newborns, adapting to the process and the family dynamics are thought to be affected by this first notification period, so it is assumed to be a very important period. There have been important speculations about notifications regarding the child's physical and psychological well-being and about the parent's unhappiness. It was found that there is insufficient literature about how and when the parents should be notified about their child's handicap; in fact, it was noticed that this topic was not covered in our country at all. In this light, the aim of this study is to determine how mothers who have children with Down's syndrome are notified, and how they prefer to be notified. **Material and Method:** In order to determine the mothers' preferences for the notification, a comprehensive literature survey, and interviews that are a qualitative method have been used. The participants consisted of 21 mothers whose children attended two different private rehabilitation centers in İstanbul and Karabük. In analyzing the data, the "Interview Form" and the transcripts of the interview recordings were used. Consecutive coding from A1 to A21 was done to describe the participants. **Results:** The findings show that the mothers were not pleased with the first notifications and that they are critical of the current notification system. According to our results, various suggestions to doctors were done. **Discussion:** Informing mothers that they have a different child is an important task for doctors that will affect the relationship between this child and the mother throughout their lives. For this reason, it is very important that doctors are knowledgeable and sensitive about the reporting process.

Keywords

Down Syndrome; Parents; Diagnose; Acceptance

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Introduction

Down's syndrome was the first chromosomal disease defined in the literature. It was defined by J. Langdon Down in 1866. In 1959, Lejeune, Gautier, and Turpin proved that the reason of the disease is the extra numbers of chromosomes. Resulting from the trisomy of the 21st chromosome, Down's syndrome can occur in 1 out of 700 births and can be diagnosed during pregnancy [6]. These children may have phenotypic conditions as well as a deficiency in growth and cognition and also many health problems in different body systems [2]. From their birth onwards, they need to receive care and support to cope with these difficult conditions.

All parents would like to have a completely healthy child. "To have a special child" is experienced differently in all families and for some, it can be more painful and hard. They want to employ positive methods to handle or their coping skills may fade. As a result, they can experience physical and emotional burnout syndrome, psychosomatic illnesses, social isolation, drug addiction, inappropriate anger problems, ignorance for the child's illness and they might keep away from the child, wearing themselves and the children out [9]. For this reason, the first approach while giving information to the parents about the medical condition of the child and the statements used are very important for the process which follows.

Informing mothers of disabled children can be seen as delivering bad news and the role of healthcare professionals is very important. After the diagnosis, healthcare professionals will be faced with various questions such as: "How to explain the diagnosis? What are some appropriate responses? How to deliver the news in a birth environment? How should the situation be discussed with the other parents informed about the birth?" It is very important that mothers are provided with the correct approach in the first process of learning about the syndrome that is thought to affect many areas such as acceptance of the newborn, adapting to the process and family dynamics. The timing of giving the news to the mother, choosing "the correct words" and up-to-date information are very crucial. According to researches, the positive approaches of healthcare professionals adopted towards the mother might determine the reaction of the mother to this unexpected situation. However, when there is a doubt about the disability of the baby or it needs confirmation, healthcare professionals feel disturbed about giving information or are unprepared. Doctors might refrain from giving information to the parents about the ultimate diagnosis. In the literature, it is reported that birth centers have made protocols on this subject, it is standing out that there is not enough study on how and when to give information to the parents about the physical or mental disabilities of the child and in our country, there is not any research on this topic [10].

This study aims to explain how the mothers learn about the Down's Syndrome diagnosis, what they learn about the diagnosis and their choice of being informed; giving a draft to healthcare professionals on how to communicate and inform the parents about such a delicate subject and give information to parents both before and after birth.

Accompanying a sensitive and appropriate treatment, the sincere and affectionate approach of healthcare professionals will help the family accept the fact and assist the parents of the disabled child in this lifelong journey.

Material and Methods

This research is structured by using qualitative research methods and techniques. Qualitative research is a process of making meaning by questioning social life and human problems with their own methods [3]. In the qualitative research process, the researcher presents the integrated research table, uses word analysis, detailed participant interview reports, and organizes the research in a natural setting. The qualitative research process is generally followed from part to entire /whole (inductive). In general terms, the qualitative researcher maintains the process by explaining concepts, meanings, and relationships, starting from observations, interviews and documents [5]. In this study, the semi-structured interview model was used as a qualitative research model.

Population and Sampling:

The population of the research was formed of 21 mothers having child with Down syndrome in two separate Special Education and Rehabilitation Centers training from different states which are Karabük and İstanbul. The average age of the mothers is 42.3 and their ages range from 28 to 63 years. The average age of children is 8.3, the oldest being 25 and the youngest being 1 year. There are six mothers who graduated from high schools, and six mothers from primary schools, five mothers from high schools, and three mothers from junior high schools. Lastly, one mother was not literate.

Collection of Data:

The data of the study were collected using the semi-structure. The semi-structured interview form was developed by researchers, taking into account similar studies in the literature. The form contains demographic information and 9 open-ended questions (Table 1.). The boundary of interview topics and questionnaires were defined and the interviews were recorded in this framework by recording with a voice recorder. Each interview lasted approximately 20 minutes, at least six minutes and maximum 73 minutes in accordance with the intended purpose and in the interactive environment. Mothers who agreed to participate in the interviews organized at the Special Education and Rehabilitation Centers were interviewed at the time of their child's education at the institution. Interviews were organized in an empty room provided by the institution. Mothers who participated in the research were given a "consent form" to explain the purpose of study and to approve their participation.

Table 1. Semi-structured Interview Form

1. When did you know your child has Down's syndrome? (before birth, after birth)
2. Who, where, when, and how did this news come to you?
3. What happened to your first reaction after learning the news? What do you think? What did you feel?
4. With whom did you share after learning the news? What was their reaction?
5. Was the initial information about Down Syndrome which is given to you sufficient?
6. How have you survived the process of accepting your child having Down Syndrome?
What helped you accept your child's diagnosis of Down's syndrome
7. Is it advantageous or disadvantageous to learn after birth? or Would you like to know this news before birth? Did you learn something changed (in terms of termination of pregnancy)
8. Which person directed you with special education? And when?
9. How would you recommend the families who are in your situation to be informed about this news? (who, where, when, how)

Analysis of Data

In the analysis of the data, "Interview Form" and written records of the data were used. These were obtained by recording with a voice recorder. Sequential coding was performed from A1 to A21 for identifying the participants. The analysis process was modeled by Marshall and Rossman (1999) in five models: a) Organizing the data; b) Definition of themes and categories; c) Testing the emerging hypothesis with data; d) the search for alternative explanations of the data and e) writing the report. In this context, when creating themes, titles can be gathered under the same heading, and titles are arranged by making quotations from appropriate speeches.

Results

The Time of Learning the Down's syndrome Diagnosis of the Children;

When the answers of the mothers to the question about when they had learnt about the Down's syndrome diagnosis of their children are investigated, it can be seen that they learnt it in time ranging from 3 days to 6 months after the birth (Table 2). Mothers expressed that they were relieved upon receiving the news. Two participants told that waiting for the final diagnosis was much more stressful.

Table 2. The Table Time of Learning the Down's Syndrome Diagnosis on The Children

Category	Participants	Frequency
Before the Birth	A3, A13, A15, A18, A19	5
On the Day The Child is Born	A4, A5, A7, A10, A11, A12, A17, A20	8
3 days- 2 months after the Birth	A1, A2, A6, A8, A9, A14, A16, A21	8

A3: "For three months, I had such a doubt which made me even worse. Isn't it so? Uncertainty is really bad. I would constantly search for children with Down's syndrome online. When my husband told me that our child has the syndrome, a huge burden was lifted off my shoulders. I calmed down, I settled down."

A5: "I lived with that suspicion for 6 months. Then, it took one more month before it became certain. For 6 months, I prayed for it not to happen. Waiting for 7 months was worse than learning about it."

A18 stated that learning about the syndrome before the birth was very important in order to be prepared for it: "It is an advantage to learn about the syndrome before birth. At least, one gets used to the idea and prepares herself for the possibility. I no longer pray for the syndrome not to occur, sure. If I had learnt about it after giving the birth, the shocking effect could have lasted longer. It could have been much more difficult to accept. Facing the fact all of a sudden while expecting a normal child would be harder."

A14 who said that she could have had an abortion: "To tell the truth, I would have had abortion thinking about the difficulties and distress that my child endured from his family life to school performance. I would not do it for myself, thinking about my ego or for feeling comfortable, but for my child so as not to make him endure such a difficulty".

Our participant A21 argues that not knowing about the syndrome increases anxiety and sadness. Learning about the disease makes the hard period of pregnancy more difficult: "I think that learning about the syndrome after giving birth contributed

to my beautiful period of pregnancy. Luckily, I learnt it after the birth because I do not know what I would do if I learnt it before. I would not know how to cope with it, my pregnancy would be painful. Fortunately, I learnt it after the birth."

Nine mothers who learnt about the first suspicion of the child after the birth got the information from the pediatrician, two of got them from the gynecologist, two of them got the information from the explanation of both the pediatrician and the gynecologist; two mothers had a suspicion from their prior knowledge about the symptoms (Table 2).

The evaluation of how and from whom the mothers learnt that their children had Down's syndrome:

Participant mothers explained their negative experiences with regard to the question: "Who gave you the information that your child has Down's syndrome and how?"

A16: "The Pediatrician told me: "Your child is retarded. He will not ask for any bread, water or do anything. He will eat only what you feed him or ask nothing." This is how I learnt my child was disabled."

A11: "Our obstetrician told me the news right before he cut the umbilical cord. It was a vaginal delivery, he told me during delivery. It was awful. He told me that my child was a healthy Mongolian baby. I was devastated. I did not know what a Mongolian baby was. He told me that he would not be able to talk, walk or go to school like normal children. He told all this at the delivery room".

A19: "I no longer go to the genetics department. There was a woman whose name started with Z and she directly told me: "Why did you give birth?" When I left her room

I was like "I gave birth to a monster, why did they give it to me, what am I supposed to do with it"? When I went there for the second time, there was another professor. "You gave birth to this baby because of motherly instincts but he will suffer a lot. You did a wrong thing, why did you bring this child to this world" he said.

A21: "The information given was very insufficient. It was this ambiguity that made me uncomfortable. It was the missing information. It was a wrong information. Maybe I was not given any information because I was a doctor. However, it is very incorrect. The person who is explaining should explain the facts with great care and detail as if the person who he/she is talking to is not a professional and knows about nothing".

Table 3. The Table about How and from Whom the Mothers Learnt that Their Children Had Down's Syndrome

The Person who Informed	Time of Information	Participant	Frequency
Pediatrician	The first examination after birth	A4,A5, A8, A14, A17	5
Gynecologist	Pregnancy control	A3, A12, A13, A15, A18	5
Pediatrician	When they arrive at the hospital for flu, jaundice, vomiting or other health problems	A1, A2, A6, A16	4
Gynecologist	At the delivery room after normal birth	A11, A20	2
Gynecologist and Pediatrician	Before the mother gained consciousness after sectio	A7, A10	2
Self-suspicious	After birth	A9	1
Radiologist	Pregnancy control	A19	1

The evaluation of opinions about how to deliver the news to the families

When the answers provided by the mothers to the question “How would you prefer that this news is told to the families and when” were examined, it is seen that they provided a range of suggestions in parallel with their own experiences of distress (Table 4).

Table 4. Opinions about How to Deliver the News to the Families

Suggestions	Participant	Frequency
Positive aspects should be mentioned	A5, A6, A11, A12, A16, A17, A19, A20, A21	9
Father should accompany the mother	A3, A11, A12, A13, A14, A17	6
Parents need to be informed in detail about benefits and threats and all stages of development and possible health problems	A4, A7, A12, A18, A20, A21	6
It should be told after a couple of hours/ days after birth	A10, A11, A12, A13	4
The whole family should be present	A2, A4, A10	3
A psychologist should accompany while explaining	A4, A6, A10	3
It needs to be explained in a compassionate, empathetic and moderate way	A5, A11, A21	3
Pictures of other children with Down's Syndrome needs to be shown	A1, A2, A5	3
Gynecologist should inform the patient	A12, A13	2
Father needs to be informed before the mother and father should explain it to the mother	A7, A8	2

Nine participants suggested that the initial information on Down syndrome should also include the positive aspects of the disease. One of these propositions was included in the participant's statements as follows:

A5: *“They say it will be good, not bad. No need to talk about bad probability. He will be fine when I deal with him. Then I would be happier and I would not get exhausted. There is a choice of telling good news before telling bad. When doctors say bad things, you think your kid will not have a reason.”*

According to the distribution of the data, the second most frequent respondent is “the husband should be next to his wife (mother)” and as the reason for this:

A14: *“A person should not be alone when learning this news. I would very much like to share this burden with my husband. What mother and father need to do, two people who bring the child to the world, also father should do the truth consciously. The couples must be together when this news is given so that they can learn what to do.”*

Six participants expressed similarity with A7 that it is important to be informed about the development of their children and possible health problems:

“What the Down syndrome is, what features does it have, deficiencies, improvements, developments should be explained, and it should be said that there may be some discomforts after giving birth. It should be said that they can come up to a certain degree with education. Physical therapy should be recommended. I mean they need to focus on education. If they have discomfort, they should be directed to the treatment.”

The evaluation in terms of how mothers felt when they learnt

about the syndrome

Mothers were asked the following questions: “What was your first reaction after learning about the syndrome? What did you think? How did you feel?” and their answers are collected under five categories namely future anxiety, sorrow, acceptance, denial and despair. Table 5 consists of the distribution of the mothers' first feelings.

Table 5. How Mothers Felt When They Learnt about The Syndrome

First Reaction	Participant	Frequency
Future anxiety	A1, A6, A7, A8, A9, A11, A16, A17, A18, A19	10
Sorrow	A1, A6, A7, A8, A12, A13, A17	7
Acceptancy	A2, A3, A4, A10, A13, A14, A19	7
Denial	A5, A7, A8, A16, A21	5
Desperation	A11, A12, A13, A20	4

Mentioning problems about future anxiety, A8 has important expressions: *“How can I look after my baby? Will I be powerful enough? Will I have any patience? What can I offer him? People are not informed about these issues. This child is special and first we thought about what we can offer him. I was sorry. It was my first child. I got married late. I was pregnant without any treatment. We were really happy because we didn't think of another child because of my age. We hoped for the best.”*

Participant A21 says the first feelings changed during this process: *“I wanted to get an eraser and erase it or change the time. I cursed my faith. When my baby came out of the incubator, all these feelings changed. I took my baby in my arms and we made a good connection from that moment onwards.”*

The evaluation in terms of factors, which have effect on the acceptance process of mothers

Eleven participants gave answers to the question about how they got over the process of accepting the fact that their children have Down's syndrome as they believed in faith. Other participants gave information about how they coped with and accepted the situation with a range of answers. The distribution of answers is given in Table 6.

Table 6. The Factors, which have effect on the Acceptance Process of Mothers

Category	Participant	Frequency
Surrender in Faith/ God (belief)	A3, A4, A7, A10, A11, A13, A15, A16, A17, A18, A19	11
Focusing on the education of the child by limiting herself to the house	A5, A6	2
Attending trainings and getting information	A9, A20	2
Observing other children with Down's Syndrome who are better	A2, A9	2
Defending the fact that acceptancy process continues for a lifetime	A21	1

Because of their religious beliefs, they saw this as a “trial” or “grace” during the acceptance of the child.

A11: *“We thought that this is a grace from God. We always think about good things. We accepted and I recovered. I hugged her differently. I have two more children, but I can say that I have learned and experienced the feeling of motherhood with my daughter.”*

A19: *“I said one night: “My God, this human being is yours, and you*

gave the feeling of motherhood, and you gave me this child. You gave the health. If it is a punishment it's my punishment if it is a reward, it's my reward. If I deserved it, this came from you. But this person is very incapable. You lead me to an exit" I prayed."

A4: *"It lasted no more than a week. What are you going to do? God has appreciated and gave. You can not put your child outside the door, or you cannot give your child to someone else, you have to look after your child."*

Stating that she has received too much support to get through the acceptance process, A21 said she feels better when she sees that her child can do something. Unlike all participants, she asserted that the acceptance process precedes life long: *"The admission process is something that you have to face again when that child encounters everything that needs to be done. I accept it is not a completed process. You are constantly facing. When someone comes and asks you what problem does the child have, then you remember once again even if you forgot it."*

The evaluation in terms of being informed about special education

Mothers who learned about the importance of physical therapy and special education started physical education and the training process a couple of months after giving a birth with the guidance of a genetic doctor. Being different from the other candidates, A1 who learnt about special education via a radio channel expressed what she experienced 13 years ago: *"When the baby was 3-4 years old, we used to hear every announcement made listening to the radios and leant about special education back then. The doctors did not mention any education when we visited the hospital for the vaccines. We never thought that education needed to start early. We thought it needed to start at the schooling age."*

Table 7. Being Informed about Special Education

Being Informed about Special Education	Participant	Frequency
Genetics doctor advised	A3, A4, A8, A9, A10, A11, A12, A13, A14, A17, A18	11
Made research on her own	A7, A15, A16, A19, A21	5
Learnt from the radio	A1	1
One neighbor who goes to special education informed	A2	1
Told by the relative	A5	1
Pediatrician advised	A6	1
Moved to disabled center	A20	1

Discussion

After the doubt disappears and diagnosis is accurate, families are informed by an expert. An Expert should consult the family in a suitable time and context. Both of parents should take part in this interview and one of the relatives can come next to them. This information should be given with pediatricist who follow up the child should be together with them. Parents should be welcomed with a positive and sincerely facial expression. Parents should be met with a positive and intimate facial expression. Parents should not be approached with pity as if doctors were giving bad news, or on the contrary, they should not have a paternalistic sense of air. It should be kept in mind that this interview is an informative meeting. Firstly, an expert should start with brief information about what is the Down syndrome and he should talk about the positive characteristics of these

children. Afterwards, should be explained in a very brief way the medical and social difficulties life related to DS that will affect the child's. It should be guided about the importance of the early start of special education, physical therapy and the process where to be applied. In this first meeting, the questions of the family must be answered clearly and briefly and then the family should be referred to a Medical Genetics or Child Genetics Specialist for further genetic counseling. Resource books list and the names of the institutions and associations to which they can apply should be given to family. Legal and social rights should be briefed. If the child is diagnosed during pregnancy, under no circumstances should not be given to directive about he termination or continuation of pregnancy to the family.

"The doctor has a heavy rock in his hand, how can he deliver it so that the mother is not crushed under this rock. It is also possible to stay under this rock or holding this rock properly and also sculpting it."

Scientific Responsibility Statement

The authors declare that they are responsible for the article's scientific content including study design, data collection, analysis and interpretation, writing, some of the main line, or all of the preparation and scientific review of the contents and approval of the final version of the article.

Animal and human rights statement

All procedures performed in this study were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards. No animal or human studies were carried out by the authors for this article.

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References

1. Abdelmuktader AM, Abd Elhamed KA. Egyptian mothers' preferences regarding how physicians break bad news about their child's disability. A structured verbal questionnaire. BMC Med Ethics.2012;13-14. DOI: 10.1186/1472-6939-13-14.
2. Beckman PJ. Influence of selected child characteristics on stress in families of handicapped infants, Am J Intellect Dev Disabil.1983; 88 (2):150-6.
3. Creswell JW. Qualitative inquiry and research design: Choosing among five traditions. Thousand Oaks, CA: Sage Publications.1988.
4. Marshall C, Rossman GB. Designing qualitative research.3rd ed. London: Sage Publications.1999.
5. Merriam S. Qualitative research and case study applications in education. San Francisco: Jossey-Bass.1998.
6. Lejeune J, Gautier, M, Turpin R. Study of somatic chromosomes from 9 mongoloid children. CR Hebd Seances Acad Sci. 1959;248:1721-22.
7. Shepard MP, Mahon MM. Chronic conditions and the family. In P. L. Jackson & J. A. Vessey (Eds.), Primary care of the child with a chronic condition . 3rd ed. New York: Mosby; 2000.p.50-65.
8. Skotko BG, Kishnani PS, Capone GT. Prenatal diagnosis of Down syndrome: How best to deliver the news. Am J Med Genet A. 2009; 149A: 2361-67. DOI: 10.1002/ajmg.a.33082.
9. Whaley LF, Wong DL. Nursing Care of Infants and Children, 5th ed. Mosby Company, Missouri.1995; 1017-22.
10. Wright DB, Gronroos N, Burkhardtmeier J., Johnson D. Delivering the Hard News Well: Your Child Has Mental Retardation. Southern California: Diagnostic Center. 2008.

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